



## neutral lipid storage disease with myopathy

Neutral lipid storage disease with myopathy is a condition in which fats (lipids) are stored abnormally in organs and tissues throughout the body. People with this condition have muscle weakness (myopathy) due to the accumulation of fats in muscle tissue. Other features of this condition may include a fatty liver, a weakened and enlarged heart (cardiomyopathy), inflammation of the pancreas (pancreatitis), reduced thyroid activity (hypothyroidism), and type 2 diabetes mellitus (the most common form of diabetes). Signs and symptoms of neutral lipid storage disease with myopathy vary greatly among affected individuals.

### Frequency

Neutral lipid storage disease with myopathy is a rare condition; its incidence is unknown.

### Genetic Changes

Mutations in the *PNPLA2* gene cause neutral lipid storage disease with myopathy. The *PNPLA2* gene provides instructions for making an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides. Triglycerides are an important source of stored energy in cells. These fats must be broken down into simpler molecules called fatty acids before they can be used for energy.

*PNPLA2* gene mutations impair the ATGL enzyme's ability to break down triglycerides. These triglycerides then accumulate in muscle and tissues throughout the body, resulting in the signs and symptoms of neutral lipid storage disease with myopathy.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

### Other Names for This Condition

- neutral lipid storage disease without ichthyosis
- NLSDM

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Neutral lipid storage disease with myopathy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853136/>

### Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Hypothyroidism  
<https://medlineplus.gov/ency/article/000353.htm>
- MedlinePlus Encyclopedia: Type 2 Diabetes  
<https://medlineplus.gov/ency/article/000313.htm>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Hypothyroidism  
<https://medlineplus.gov/ency/article/000353.htm>
- Encyclopedia: Type 2 Diabetes  
<https://medlineplus.gov/ency/article/000313.htm>
- Health Topic: Cardiomyopathy  
<https://medlineplus.gov/cardiomyopathy.html>
- Health Topic: Lipid Metabolism Disorders  
<https://medlineplus.gov/lipidmetabolismdisorders.html>
- Health Topic: Muscle Disorders  
<https://medlineplus.gov/muscledisorders.html>
- Health Topic: Pancreatitis  
<https://medlineplus.gov/pancreatitis.html>

### Genetic and Rare Diseases Information Center

- Neutral lipid storage disease with myopathy  
<https://rarediseases.info.nih.gov/diseases/10288/neutral-lipid-storage-disease-with-myopathy>

### Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Hypothyroidism  
<https://www.niddk.nih.gov/health-information/endocrine-diseases/hypothyroidism>
- National Institute of Neurological Disorders and Stroke: Lipid Storage Diseases Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Lipid-storage-diseases-Information-Page>
- National Institute of Neurological Disorders and Stroke: Myopathy Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Myopathy-Information-Page>

### Educational Resources

- American College of Rheumatology: Metabolic Myopathies  
<http://www.rheumatology.org/I-Am-A/Patient-Caregiver/Diseases-Conditions/Metabolic-Myopathies>
- Disease InfoSearch: Neutral lipid storage disease with myopathy  
<http://www.diseaseinfosearch.org/Neutral+lipid+storage+disease+with+myopathy/5209>
- MalaCards: neutral lipid storage disease with myopathy  
[http://www.malacards.org/card/neutral\\_lipid\\_storage\\_disease\\_with\\_myopathy](http://www.malacards.org/card/neutral_lipid_storage_disease_with_myopathy)
- Orphanet: Neutral lipid storage disease  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=165](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=165)
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/msys/cardiac2.htm#nlsdm>

### Patient Support and Advocacy Resources

- American Heart Association: Cardiomyopathy  
[http://www.heart.org/HEARTORG/Conditions/More/ChildrensHealth/Pediatric-Cardiomyopathies\\_UCM\\_312219\\_Article.jsp](http://www.heart.org/HEARTORG/Conditions/More/ChildrensHealth/Pediatric-Cardiomyopathies_UCM_312219_Article.jsp)
- Muscular Dystrophy Association: Facts About Myopathies  
[https://www.mda.org/sites/default/files/publications/Facts\\_Myopathies\\_P-208.pdf](https://www.mda.org/sites/default/files/publications/Facts_Myopathies_P-208.pdf)

## ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22neutral+lipid+storage+disease+with+myopathy%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28neutral+lipid+storage+disease%29+AND+%28myopathy%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- NEUTRAL LIPID STORAGE DISEASE WITH MYOPATHY  
<http://omim.org/entry/610717>

## **Sources for This Summary**

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